CLINICAL NOTES, CASE REPORTS AND NEW INSTRUMENTS

HEMOCHROMATOSIS IN A JAPANESE

CASE REPORT

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HEMOCHROMATOSIS, or Bronze diabetes, has been described as cirrhosis of the liver, plus pigmentation of the skin, plus diabetes, the last usually occurring in the later stages, but not necessarily present. Less than one hundred cases have been reported in the literature, and only three in Japanese. The positive clinical diagnosis is based on the demonstration of hemosiderin in a piece of excised skin; the pathological diagnosis on the presence of the two characteristic pigments, hemosiderin and hemofuscin, in the liver, pancreas and other organs. This case did not have diabetes, but the pathological findings fulfilled the criteria for diagnosis required by Dr. F. B. Mallory. Clinically the diagnosis of hemochromatosis was suggested after the unusual pigmentation was noted.

CASE REPORT

Mr. D. S. Y., a 35-year-old Japanese teacher of English first came under the medical observation of Dr. W. F. Cheney in 1911 complaining of stomach trouble. I am indebted to him for the history up until 1927. The family history and marital history were not unusual. The patient had dysentery in Japan in 1902. He did not drink and there was no history of exposure to copper. He came for treatment of sour stomach, constipation, headaches and protruding hemorrhoids, and also stated that he had the habit of rumination. His appetite was good and he could swallow a "gallon of food at a time," and bring up any amount of it up to several hours after ingestion. He had been on a restricted diet and losing weight for two years. Physical examination showed him to be small, pale and poorly nourished, but there were no abnormalities of eyes, throat, neck, heart, lungs, abdomen, reflexes or skin. The stomach outline after inflation with carbon dioxid indicated dilatation. The urine was negative. A single extraction Ewald test-meal analysis revealed a total gastric acidity of 12 degrees, with no free HCl. He was put on treatment for chronic gastritis and achlorhydria which included elixir of lactopeptin which contains 18 per cent alcohol, and hemaboloids which contain 17 per cent alcohol. He improved.

He continued his medicine for nine years when he returned with the same symptoms as before which had grown worse after influenza one month previously. He had had several minor operations for bleeding hemorrhoids. He weighed less and his abdomen was somewhat tender to palpation. The gastric test meal showed 4 degrees total acidity and no free HCl. Treatment was continued and he got on fairly well for two years, but in March, 1922, he entered the hospital for removal of hemorrhoids, which was done. At that time his urine and stools showed nothing unusual, and the Wassermann reaction was negative. The blood count was 3,470,000 red blood cells and 3,250 white blood cells. The hemoglobin was 67 per cent. A fractional gastric test meal showed no free HCl in any of the specimens and a very low total acidity. An x-ray of the gastro-intestinal tract was reported normal except for adhesions about the cecum. Just three years later he again sought medical advice, as he had been vomiting small amounts of bright blood. The physical examination was the same as previously, but the stool was strongly positive for occult blood. The blood count showed a moderate anemia with 5000 white blood cells and normal differential count. A gastro-intestinal x-ray examination did not reveal an ulcer or neoplasm and fluoroscopy of the chest was negative. In view of the hematemesis, cirrhosis of the liver was suspected although the organ seemed normal to physical examination, and the only history of alcoholic intake was that contained in the medicine prescribed.

In October, 1926, he first noted swelling of the legs, and examination showed a very large, smooth, tender liver, edema of the legs, but no signs of ascites. He had only 60 per cent hemoglobin and 2,510,000 red blood cells. A differential white blood count showed 55 per cent mononuclear cells. After three months the edema had increased involving the genitalia and there were signs of moderate ascites, but no distended veins over the abdomen. A gastro-intestinal x-ray series and a barium enema were negative. A flat plate of the abdomen showed an upper abdominal shadow interpreted as due to an enlarged liver. His blood had improved. A month afterward he became semiconscious and was sent to the hospital in January, 1927, where he died three days later. Examination while in the hospital showed an intense chestnut-brown pigmentation of a dry scaly skin, especially over the neck, lower abdomen and shins, and about the mammary glands, genitalia, and anus. Irregular pigmentation over the chest gave it a pronounced speckled appearance. He was emaciated and dehydrated and the sclerae and mucous membranes were icteric. There were many external hemorrhoids about the anus. The blood pressure was 120 systolic, 70 diastolic. Fifteen hundred and sixty cc. of abdominal ascitic fluid was removed, but neither the liver edge nor the spleen could be felt. The blood count was 2,380,000 red blood cells, 13,900 white blood cells with 86 per cent polymorphonuclear leukocytes. The hemoglobin was 68 per cent, giving a color index of 1.4. The urine was negative for sugar, but showed a faint trace of albumin with numerous hyaline casts. The blood Wassermann was negative, the blood sugar was 50 mgs. per 100 cc. of blood, the blood urea was 61.5 mgs. per 100 cc. of blood. The direct Van den Bergh reaction was strongly positive in three minutes, the indirect equaled 25 units. The icterus index was 80. The ascitic fluid was turbid, orange colored, and the specific gravity was 1018. The albumin was 4.5 grams per liter. The cells were 78 per cent polymorphonuclear leukocytes and 22 per cent mononuclears. On the third day he developed râles at both lung bases and suddenly expired.

A complete autopsy was performed. The anatomical diagnosis was hemochromatosis, chronic atrophic cirrhosis of the liver, chronic gastritis with multiple polyps of the stomach, terminal bronchopneumonia and embolism of the right pulmonary artery with infarction of the lower lobe of the right lung. The liver was very small and nodular and on cut surface and routine microscopic examination had the characteristics of the type of cirrhosis described by Laennec. The pancreas and adrenals did not show any unusual sclerosis. Microscopic sections stained for hemosiderin and hemofuscin showed large amounts of hemosiderin present in the liver and pancreas with smaller amounts in other organs. It was demonstrated in the zona glomerulosa of the adrenals. Mallory regards this finding as pathognomonic of hemochromatosis. This pigment was also present in small quantities in the coil glands of the skin from the lower leg. Hemofuscin was present in small amounts in the liver and pancreas, but was abundant in the muscularis mucosae of the stomach. Elsewhere it was present in very small amounts.

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